Johns Hopkins University
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December 16, 2004

Introduction to the Unified Medical Language System

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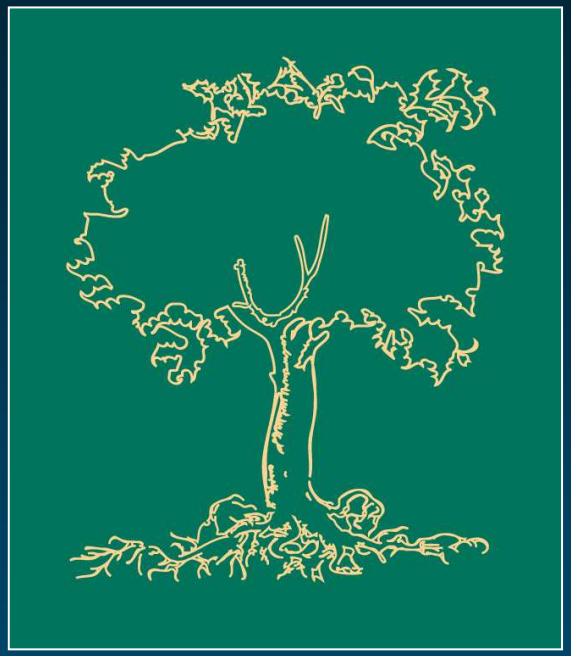
Outline

- Introduction
- Overview through an example
- The three UMLS Knowledge Sources
  - UMLS Metathesaurus
  - UMLS Semantic Network
  - SPECIALIST Lexicon and lexical tools
- UMLS in action: *MetaMap*

Introduction

What does UMLS stand for?

- Unified
- Medical
- Language
- System



UMLS®
Unified Medical Language System®
UMLS Metathesaurus®

Motivation

- Started in 1986
- National Library of Medicine
- “Long-term R&D project”
- Complementary to IAIMS

(Integrated Academic Information Management Systems)

«[…] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»

The UMLS in practice

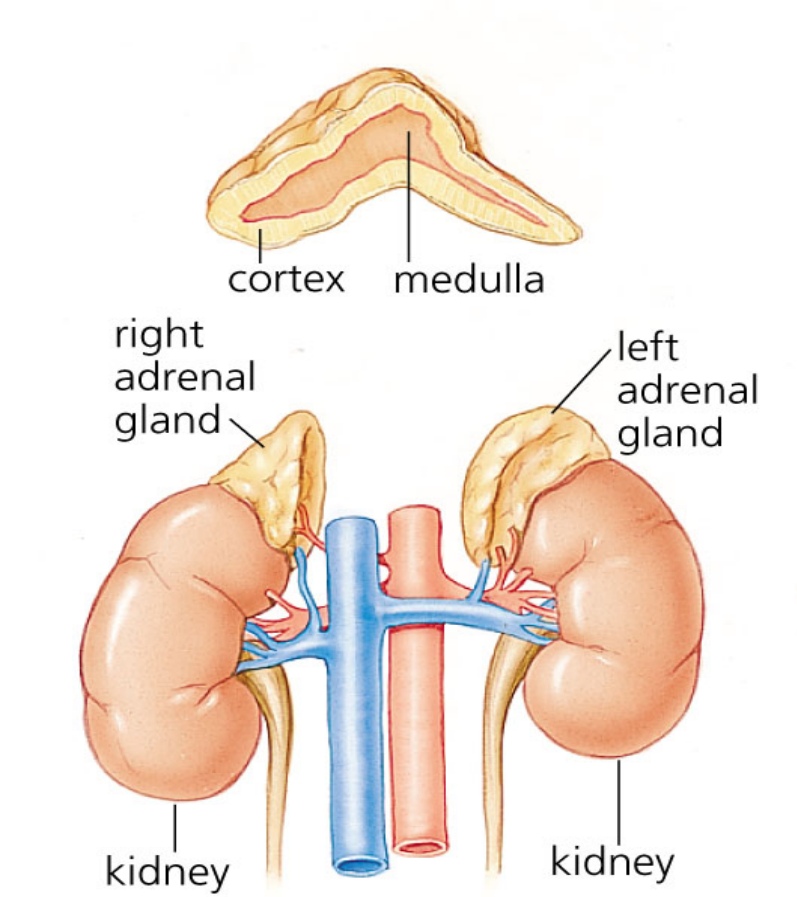
- Database
  - Series of relational files
- Interfaces
  - Web interface: Knowledge Source Server (UMLSKS)
  - Application programming interfaces (Java and XML-based)
- Applications
  - lvg (lexical programs)
  - MetamorphoSys (installation and customization)

The UMLS is *not* an end-user application

Overview through an example

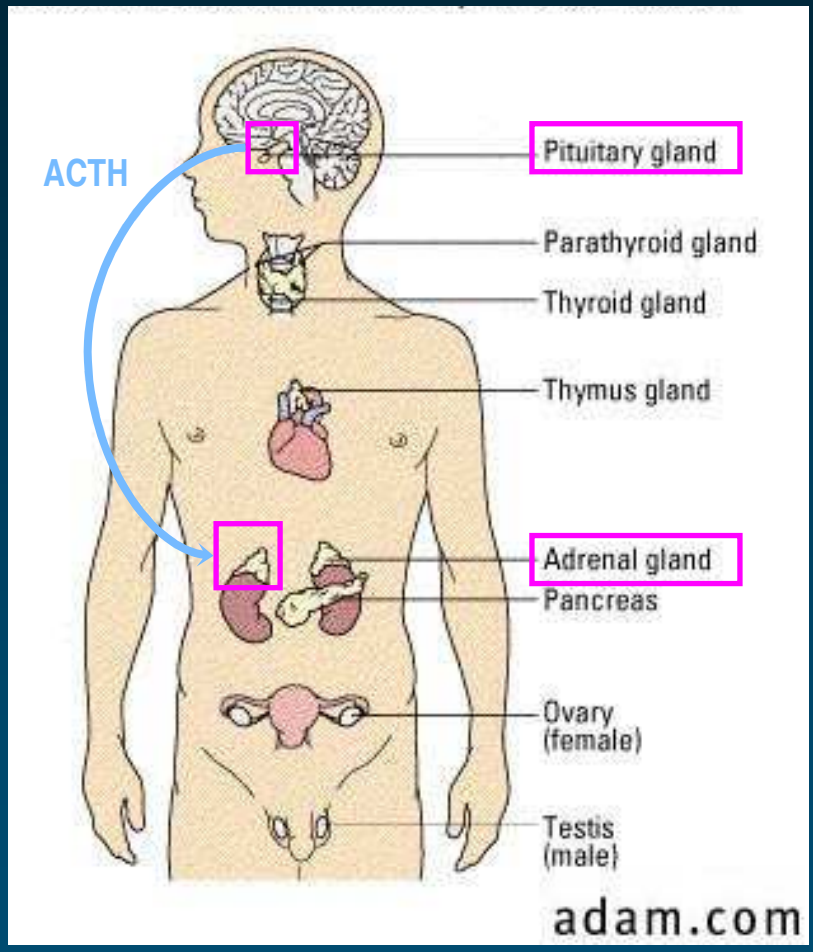
Addison’s disease

- Addison's disease is a rare endocrine disorder
- Addison's disease occurs when the adrenal glands do not produce enough of the hormone cortisol
- For this reason, the disease is sometimes called chronic adrenal insufficiency, or hypocortisolism



Adrenal insufficiency Clinical variants

- Primary / Secondary
  - Primary: lesion of the adrenal glands themselves
  - Secondary: inadequate secretion of ACTH by the pituitary gland
- Acute / Chronic
- Isolated / Polyendocrine deficiency syndrome



Addison's disease: Symptoms

- Fatigue
- Weakness
- Low blood pressure
- Pigmentation of the skin (exposed and non-exposed parts of the body)
- …

AD in medical vocabularies

- Synonyms: different terms
  - Addisonian syndrome — eponym
  - Bronzed disease — symptoms
  - Addison melanoderma — symptoms
  - Asthenia pigmentosa — symptoms
  - Primary adrenal deficiency — clinical variants
  - Primary adrenal insufficiency — clinical variants
  - Primary adrenocortical insufficiency — clinical variants
  - Chronic adrenocortical insufficiency — clinical variants
- Contexts: different hierarchies

Organize terms

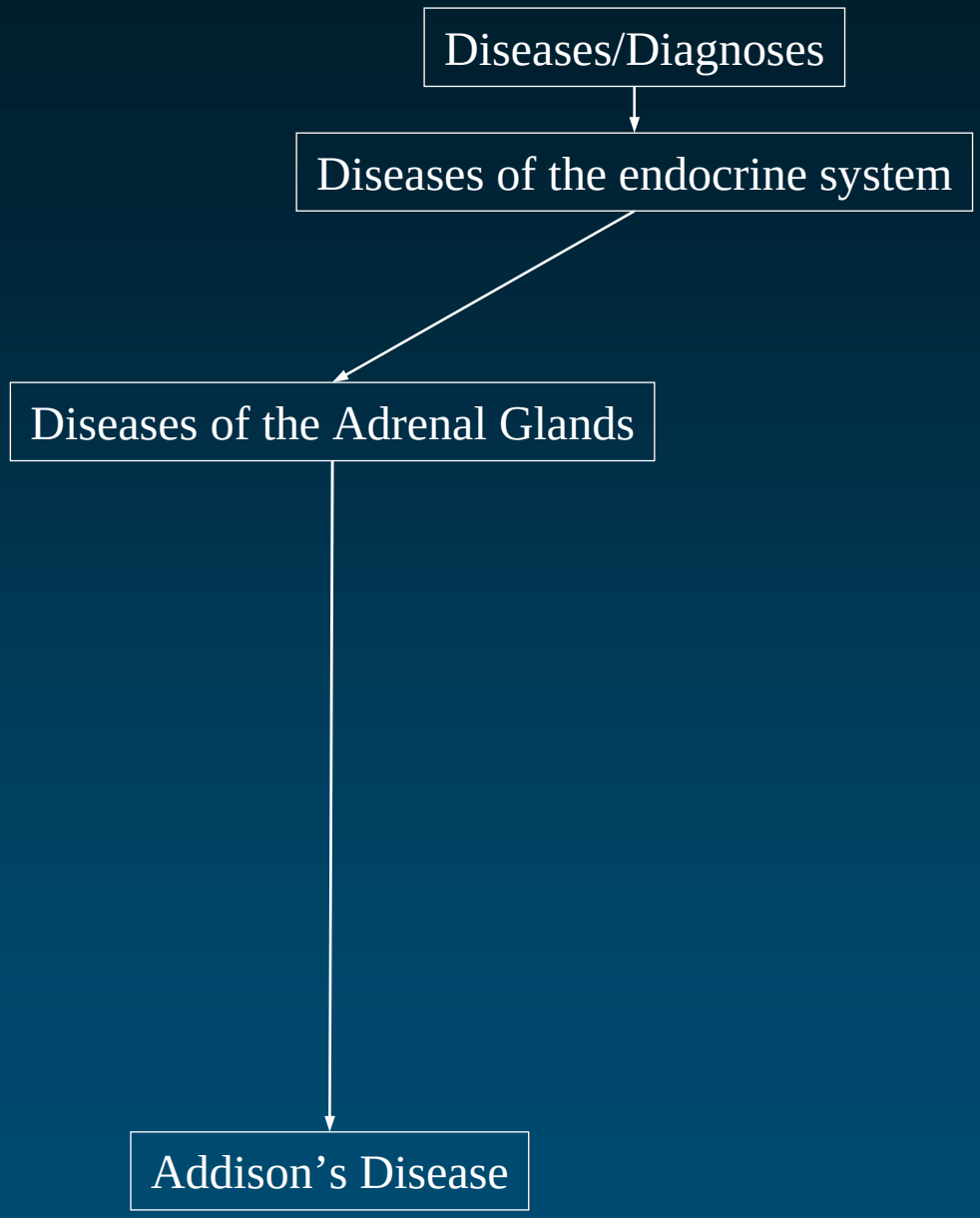
- Synonymous terms clustered into a concept
- Preferred term
- Unique identifier (CUI)

Adrenal gland diseases	MeSH	D000307
Adrenal disorder	AOD	0000005418
Disorder of adrenal gland	Read	C15z.
Diseases of the adrenal glands	SNOMED	DB-70000

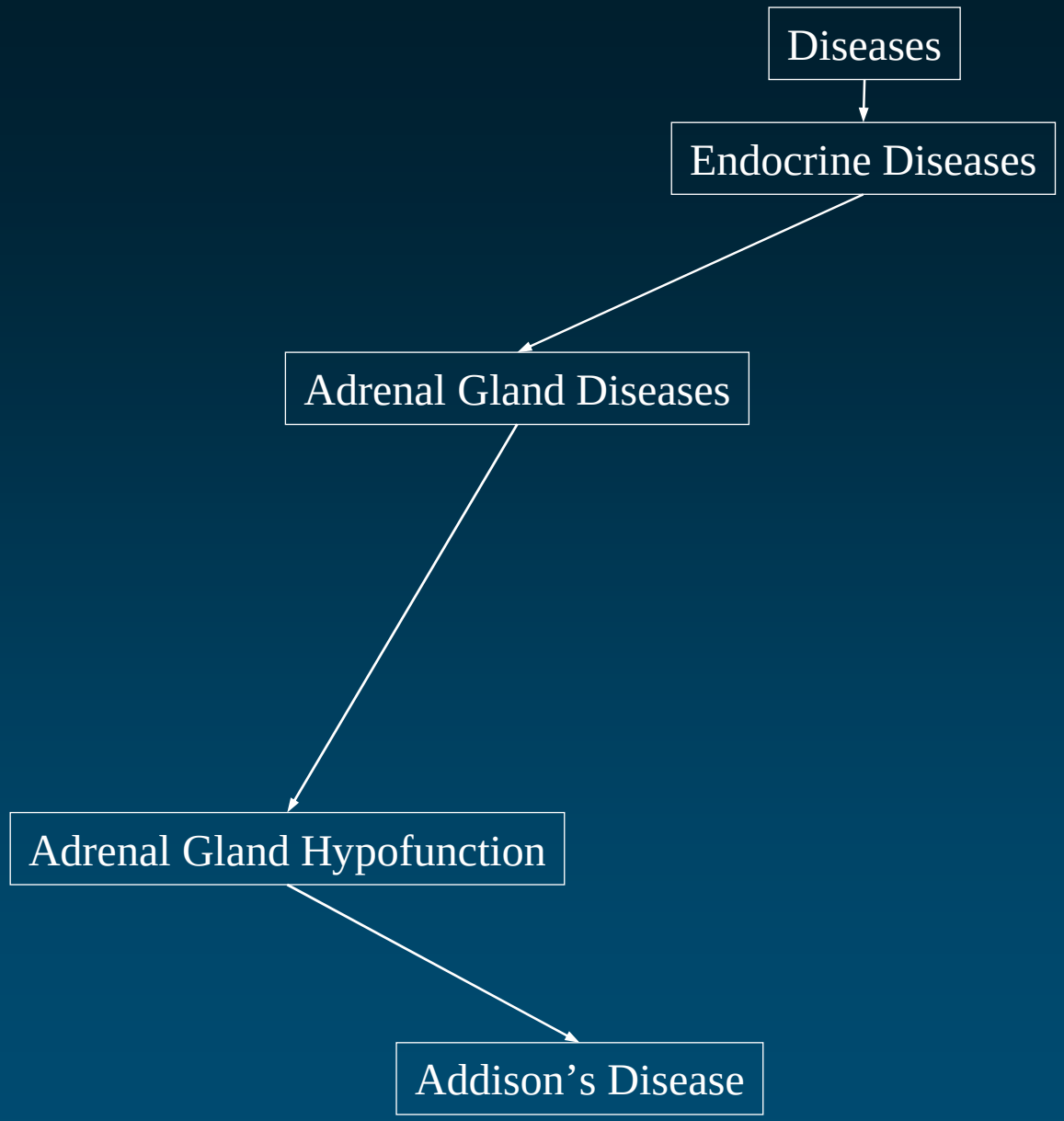
C0001621

Adrenal Gland Diseases

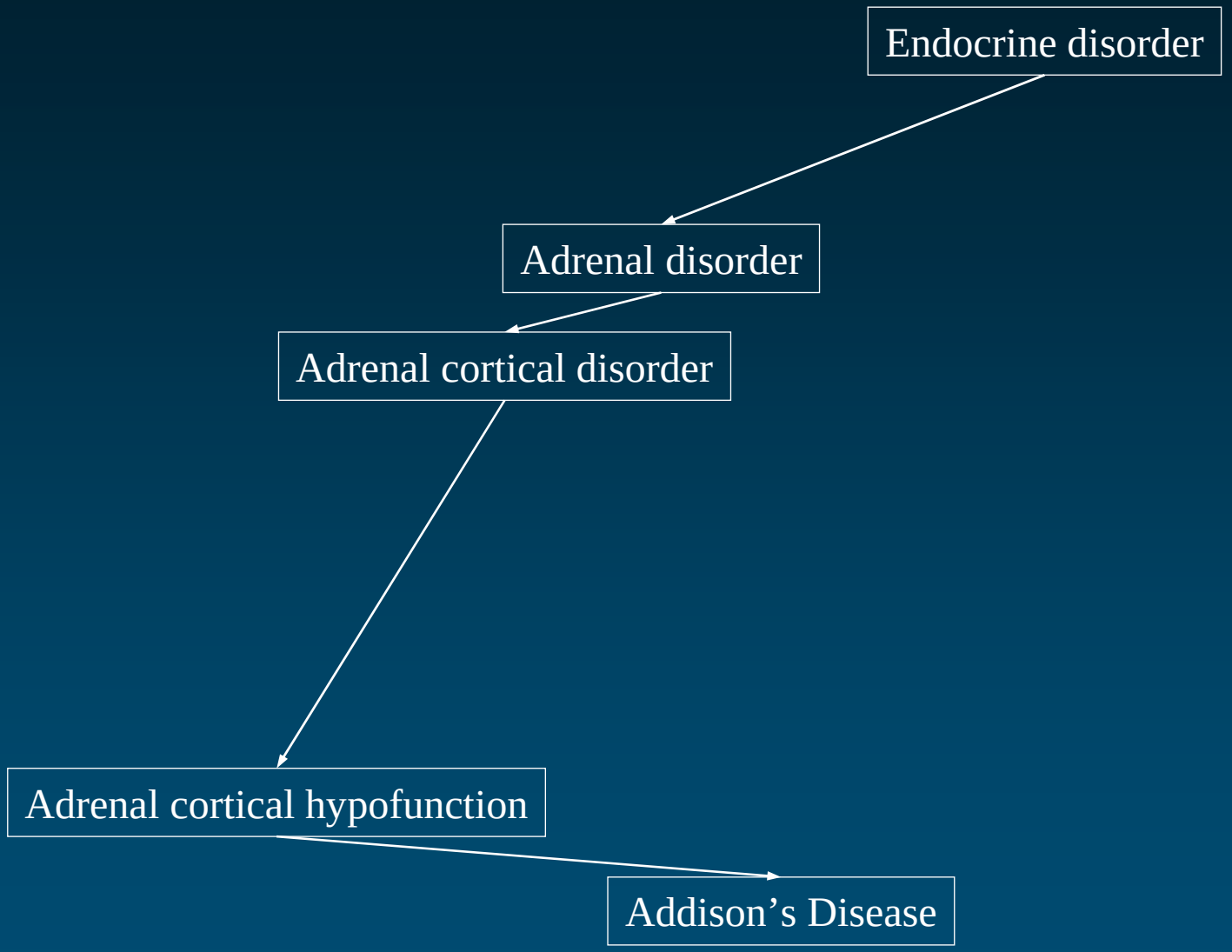
SNOMED International



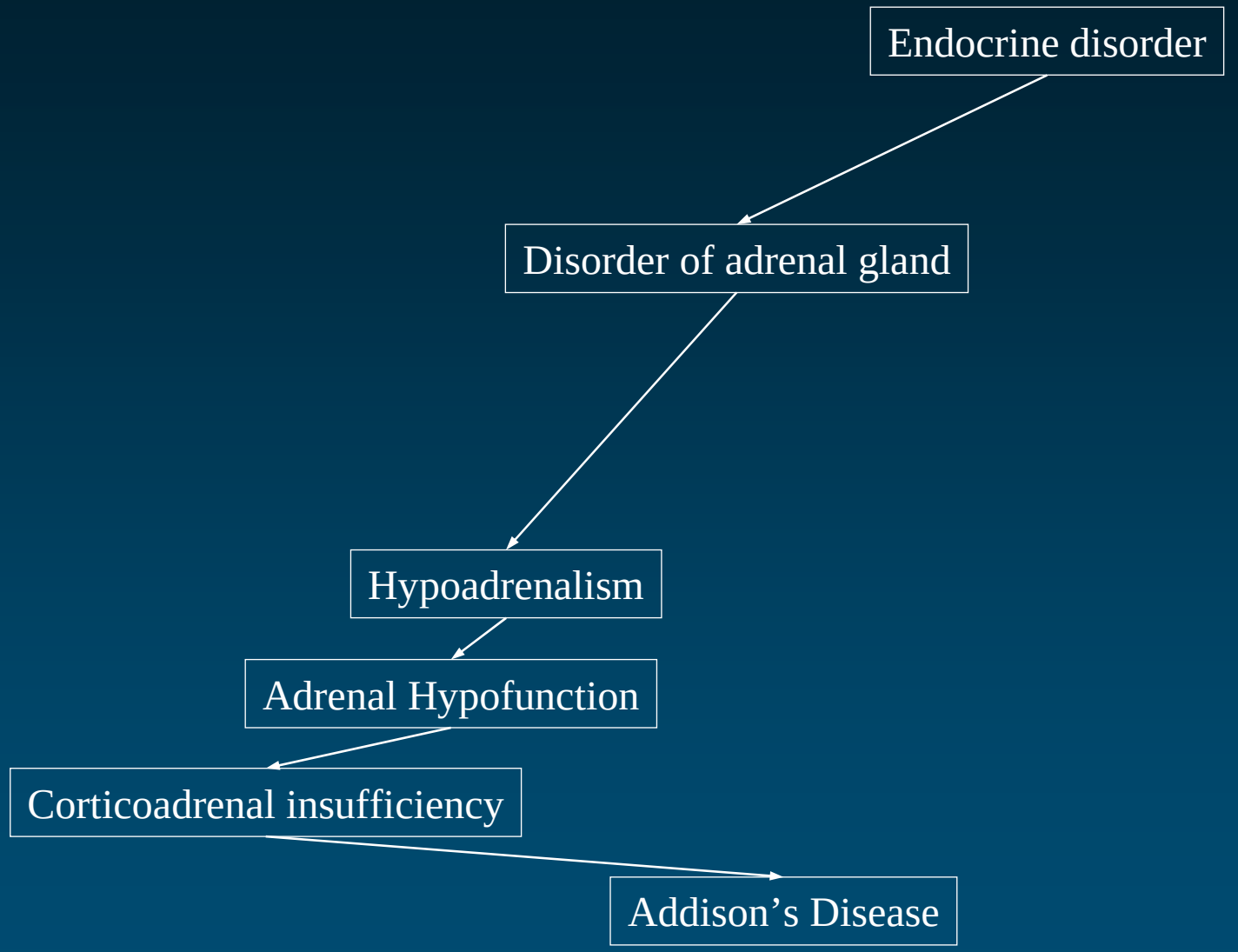
MeSH


Diseases
Endocrine Diseases
Adrenal Gland Diseases
Adrenal Gland Hypofunction
Addison’s Disease

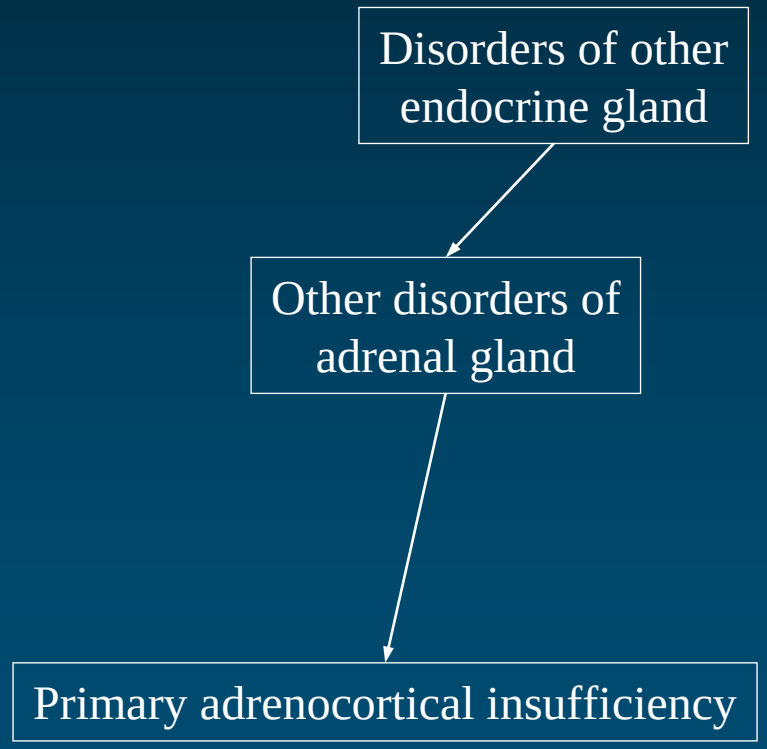
AOD


Endocrine disorder
Adrenal disorder
Adrenal cortical disorder
Adrenal cortical hypofunction
Addison’s Disease

Read Codes

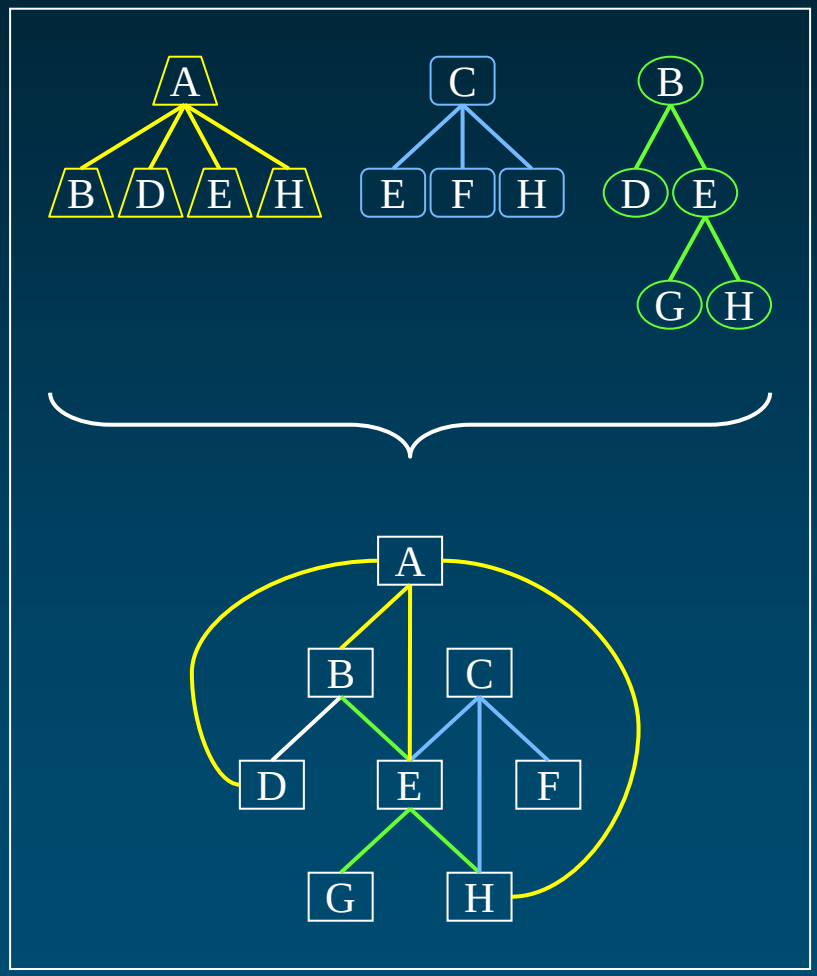

Endocrine disorder
Disorder of adrenal gland
Hypoadrenalism
Adrenal Hypofunction
Corticoadrenal insufficiency
Addison’s Disease

ICD-10


Disorders of other endocrine gland
Other disorders of adrenal gland
Primary adrenocortical insufficiency

Organize concepts

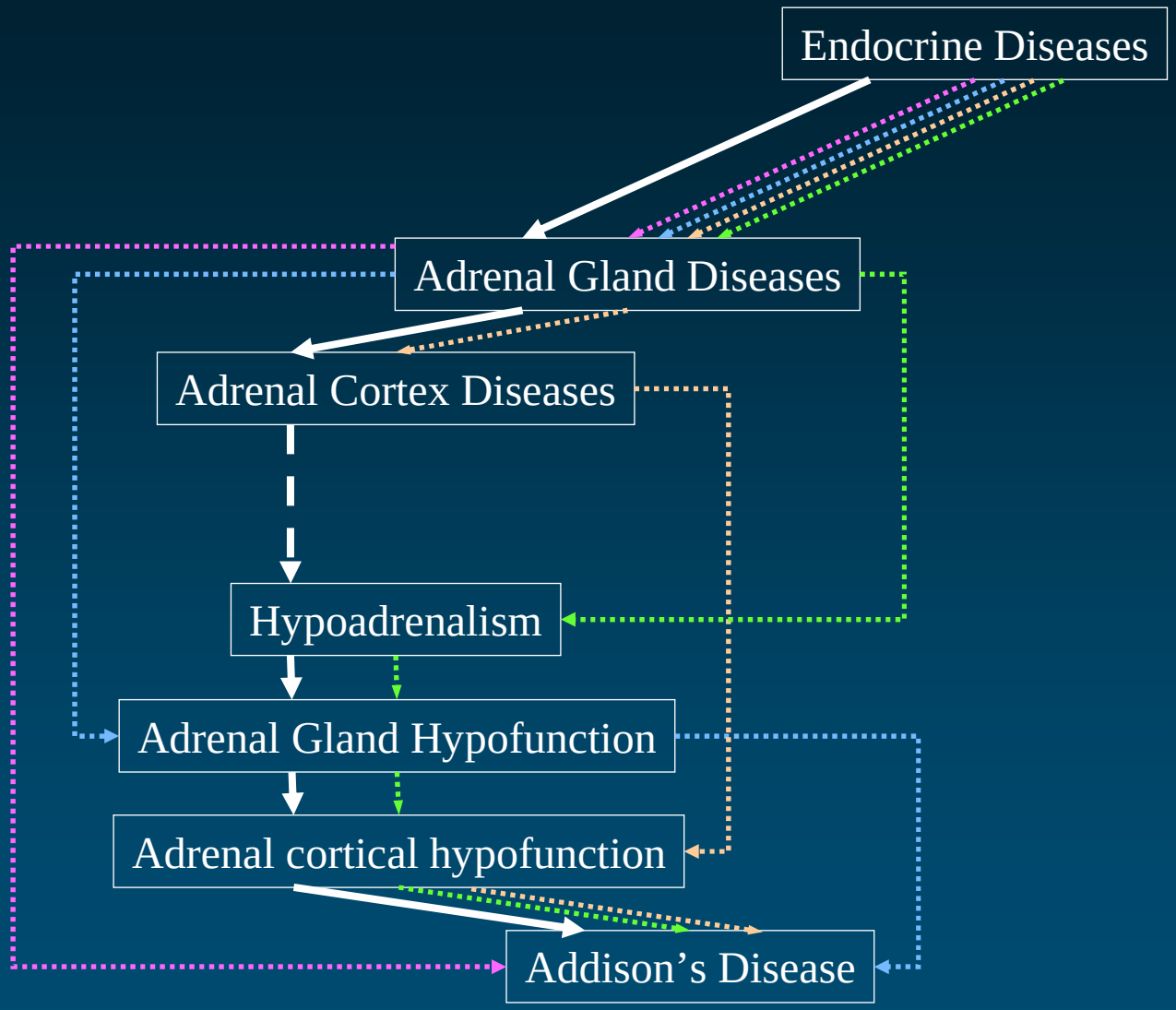
- Inter-concept relationships: hierarchies from the source vocabularies
- Redundancy: multiple paths
- One graph instead of multiple trees (multiple inheritance)



organize concepts

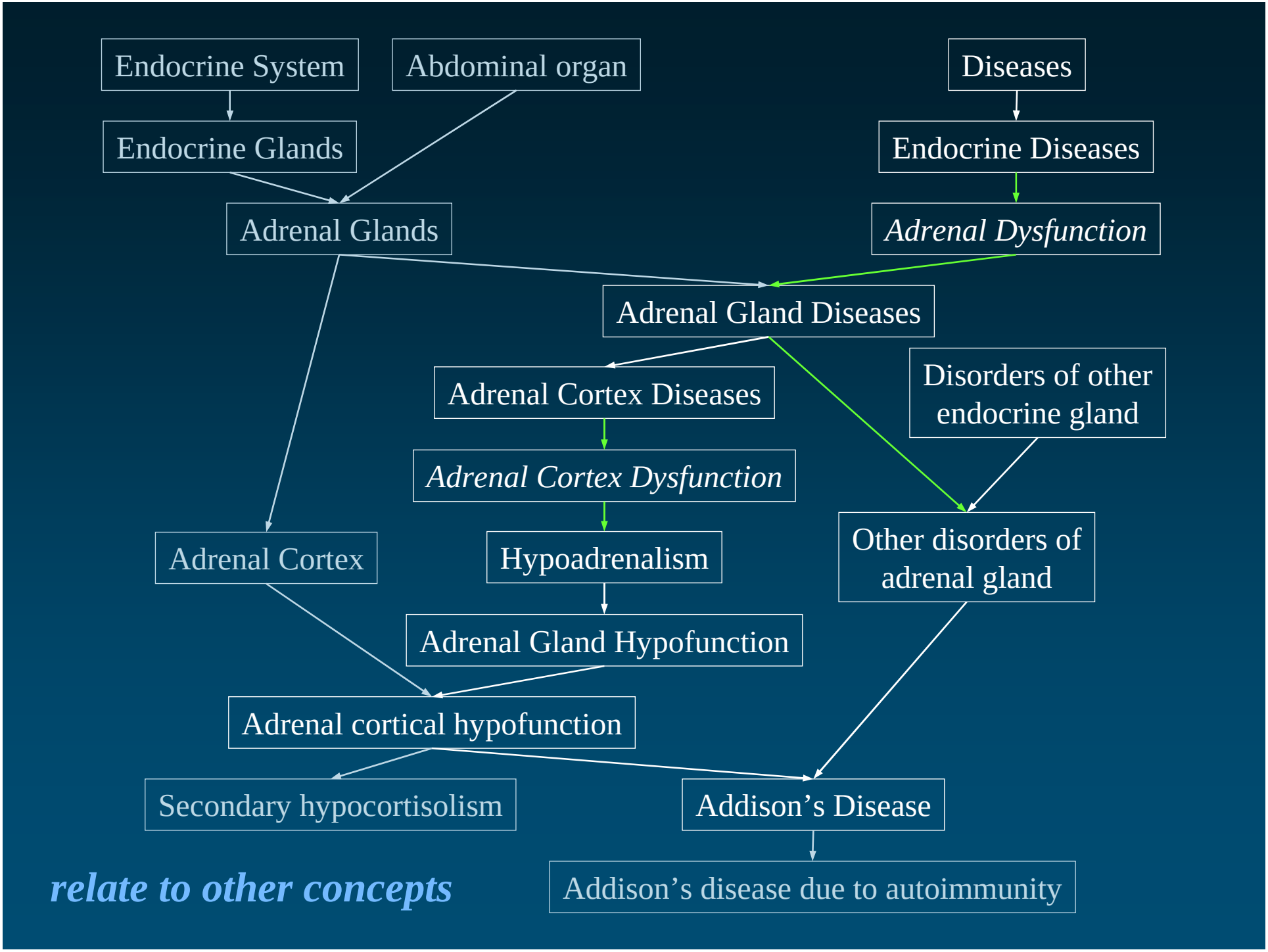
SNOMED
MeSH
AOD
Read Codes

UMLS



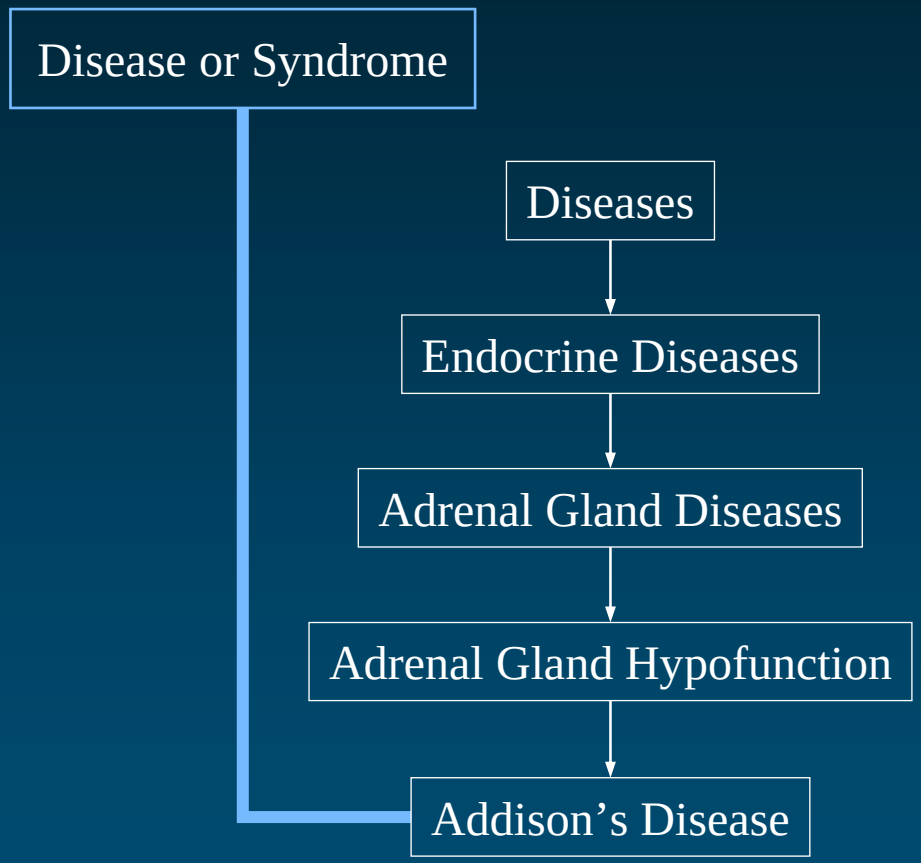
Relate to other concepts

- Additional hierarchical relationships
  - link to other trees
  - make relationships explicit
- Non-hierarchical relationships
- Co-occurring concepts
- Mapping relationships


Endocrine System
Abdominal organ
Diseases
Endocrine Glands
Endocrine Diseases
Adrenal Glands
Adrenal Dysfunction
Adrenal Gland Diseases
Adrenal Cortex Diseases
Disorders of other endocrine gland
Adrenal Cortex Dysfunction
Adrenal Cortex
Hypoadrenalism
Other disorders of adrenal gland
Adrenal Gland Hypofunction
Adrenal cortical hypofunction
Secondary hypocortisolism
Addison's Disease
relate to other concepts
Addison's disease due to autoimmunity

Categorize concepts

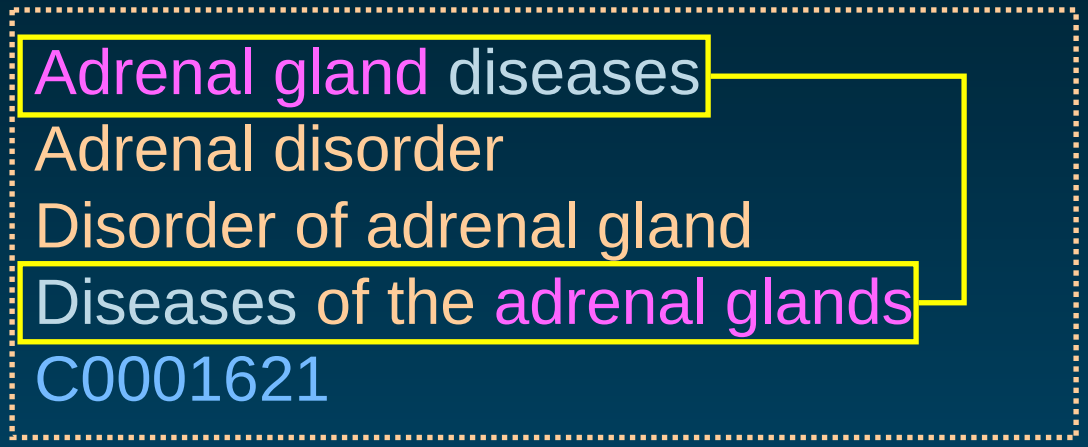
- High-level categories (semantic types)
- Assigned by the Metathesaurus editors
- Independently of the hierarchies in which these concepts are located



How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

Lexical knowledge

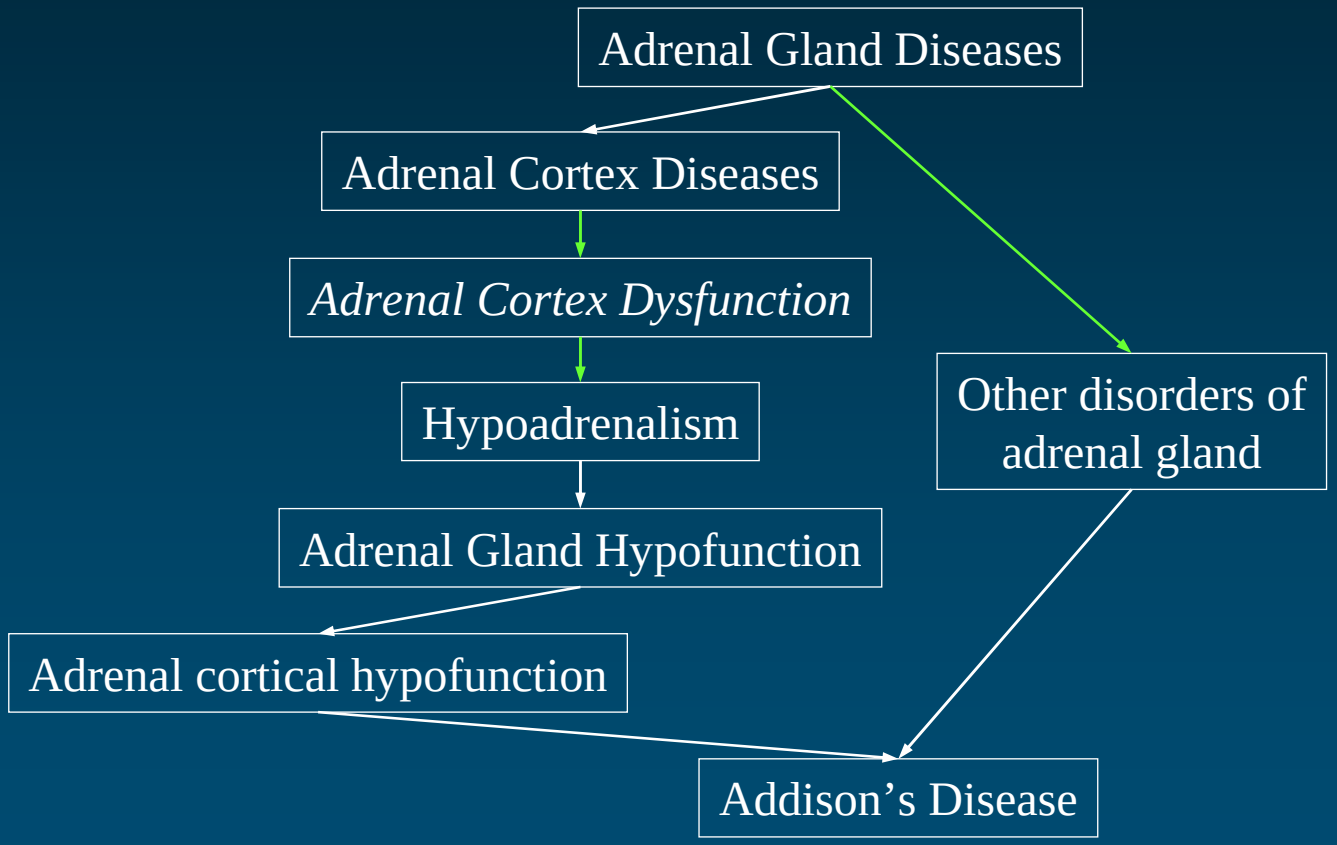


Semantic pre-processing

- Metadata in the source vocabularies

- Tentative categorization
- Positive (or negative) evidence for tentative synonymy relations based on lexical features

Additional knowledge: UMLS editors



UMLS Summary

- Synonymous terms clustered into concepts
- Unique identifier

- Finer granularity
- Broader scope
- Additional hierarchical relationships
- Semantic categorization

UMLS Knowledge Sources

UMLS 3 components

- Metathesaurus
  - Concepts
  - Inter-concept relationships
- Semantic Network
  - Semantic types
  - Semantic network relationships
- Lexical resources
  - SPECIALIST Lexicon
  - Lexical tools

UMLS Metathesaurus

Metathesaurus Basic organization

- Concepts
  - Synonymous terms are clustered into a concept
  - Properties are attached to concepts, e.g.,
    - Unique identifier
    - Definition
- Relations
  - Concepts are related to other concepts
  - Properties are attached to relations, e.g.,
    - Type of relationship
    - Source

Source Vocabularies (2004AB)

- 134 source vocabularies
  - 126 contributing concept names
- 73 families of vocabularies
  - multiple translations (e.g., MeSH, ICPC, ICD-10)
  - variants (American-English equivalents, Australian extension/adaptation)
  - subsequent editions usually considered distinct families (ICD: 9-10; DSM: IIIR-IV)
- Broad coverage of biomedicine
- Common presentation

Biomedical terminologies

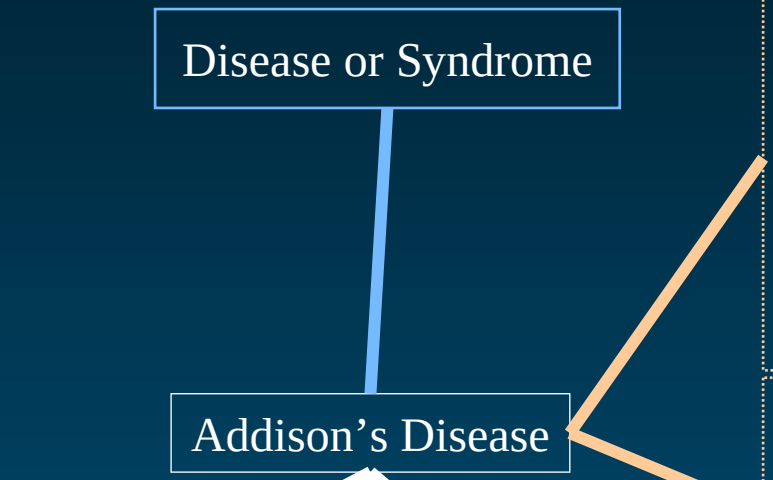
- General vocabularies
  - anatomy (UWDA, Neuronames)
  - drugs (RxNorm, First DataBank, Micromedex)
  - medical devices (UMD, SPN)
- Several perspectives
  - clinical terms (SNOMED CT)
  - information sciences (MeSH, CRISP)
  - administrative terminologies (ICD-9-CM, CPT-4)
  - data exchange terminologies (HL7, LOINC)

Biomedical terminologies (cont'd)

- Specialized vocabularies
  - nursing (NIC, NOC, NANDA, Omaha, PCDS)
  - dentistry (CDT)
  - oncology (PDQ)
  - psychiatry (DSM, APA)
  - adverse reactions (COSTART, WHO ART)
  - primary care (ICPC)
- Terminology of knowledge bases (AI/Rheum, DXplain, QMR)

The UMLS serves as a vehicle for the regulatory standards (HIPAA, CHI)

Addison's Disease: Concept



ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)
ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE
Addison melanoderma
Melasma addisonii
Primary adrenal deficiency
Asthenia pigmentosa
Bronzed disease
Insufficiency, adrenal primary
Primary adrenocortical insufficiency
Addison's, disease

MALADIE D'ADDISON - French
Addison-Krankheit - German
Morbo di Addison - Italian
DOENCA DE ADDISON - Portuguese
ADDISONOVA BOLEZN' - Russian
ENFERMEDAD DE ADDISON - Spanish

SNOMED
MeSH
AOD
Read Codes
…

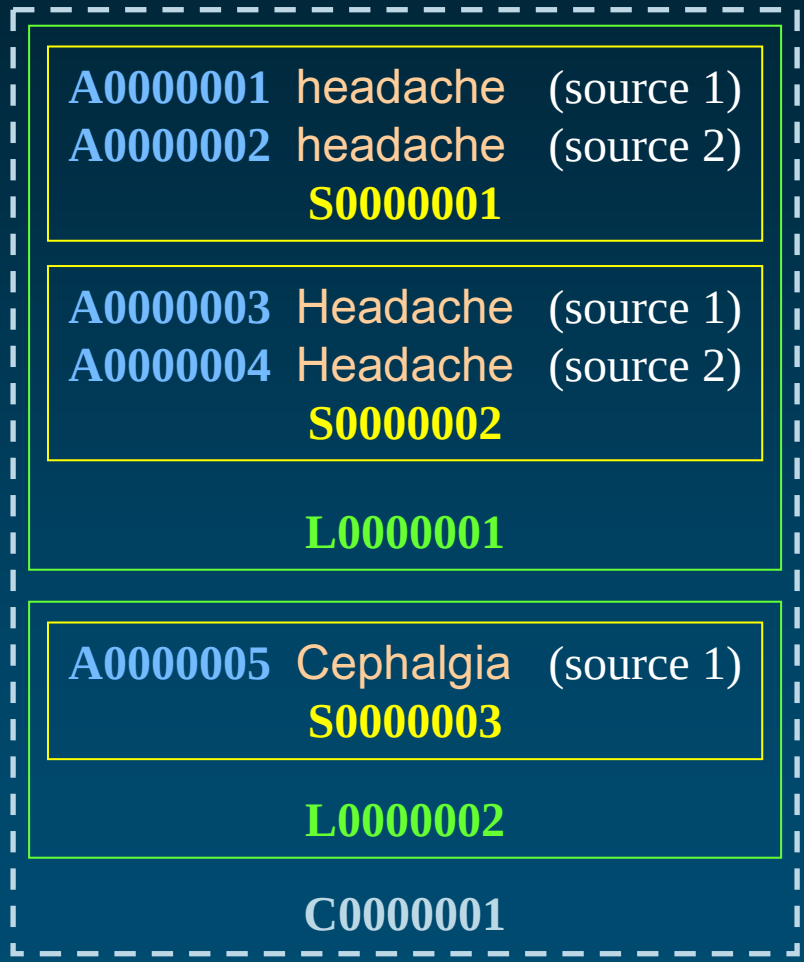
C0001403

A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal.

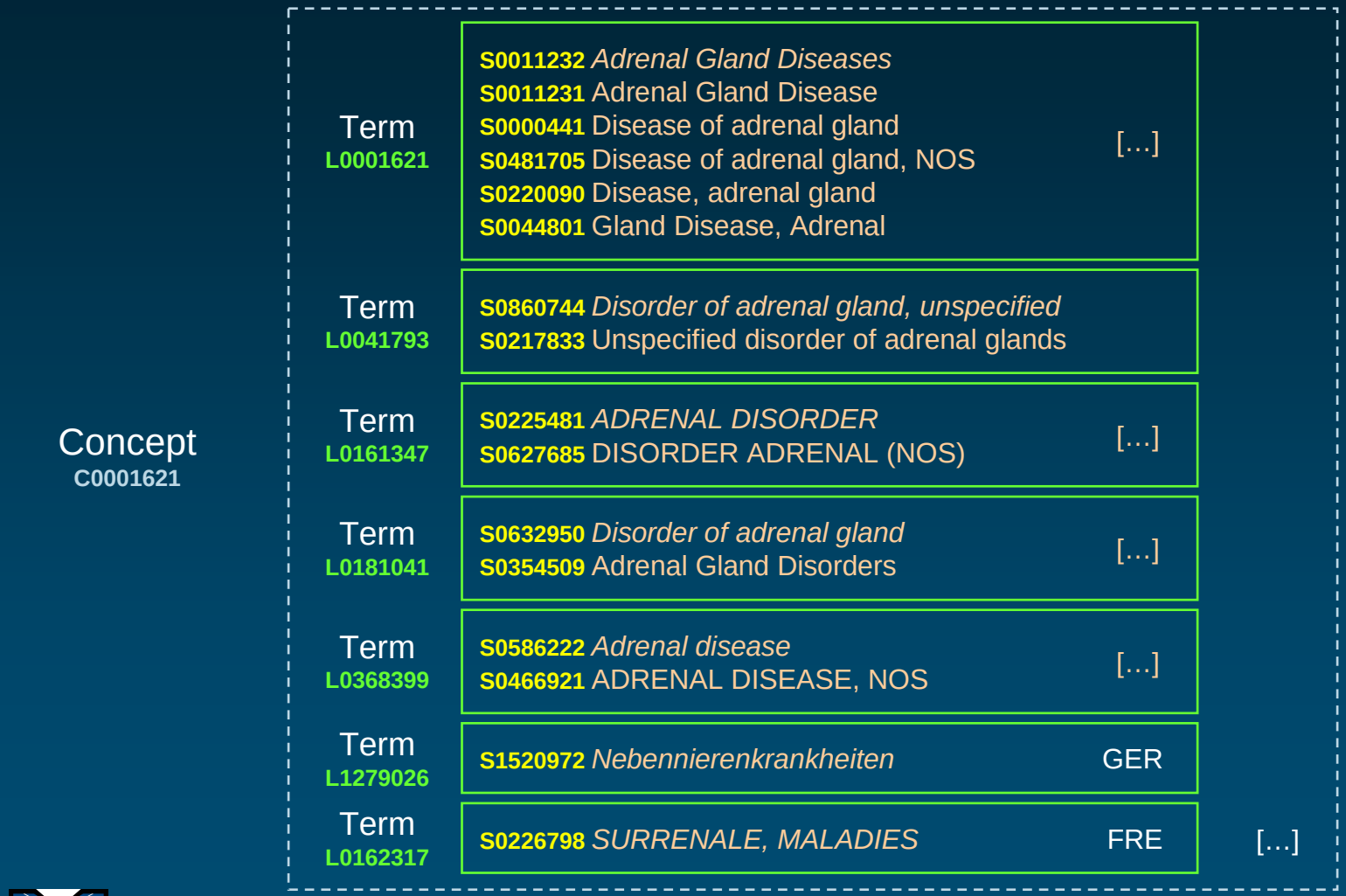
NLM

Metathesaurus Concepts (2004AB)

- Concept (> 1M) CUI
  - Set of synonymous concept names
- Term (> 3.8 M) LUI
  - Set of normalized names
- String (> 4.3M) SUI
  - Distinct concept name
- Atom (> 5.1M) AUI
  - Concept name in a given source

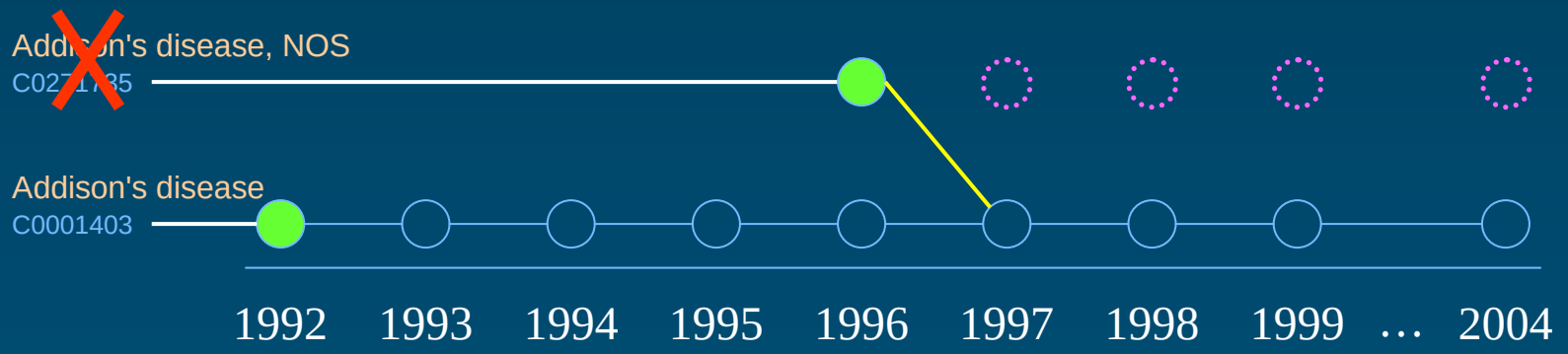


Cluster of synonymous terms



Metathesaurus Evolution over time

- Concepts never die (in principle)
  - CUIs are permanent identifiers
- What happens when they do die (in reality)?
  - Concepts can merge or split
  - Resulting in new concepts and deletions



Metathesaurus Relationships

- Symbolic relations: ~9 M pairs of concepts
- Statistical relations : ~7 M pairs of concepts (co-occurring concepts)
- Mapping relations: 100,000 pairs of concepts

- Categorization: Relationships between concepts and semantic types from the Semantic Network

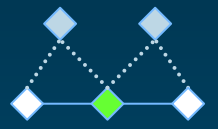
Symbolic relations

- Relation
  - Pair of “atom” identifiers
  - Type
  - Attribute (if any)
  - List of sources (for type and attribute)
- Semantics of the relationship: defined by its type [and attribute]

Source transparency: the information is recorded at the “atom” level

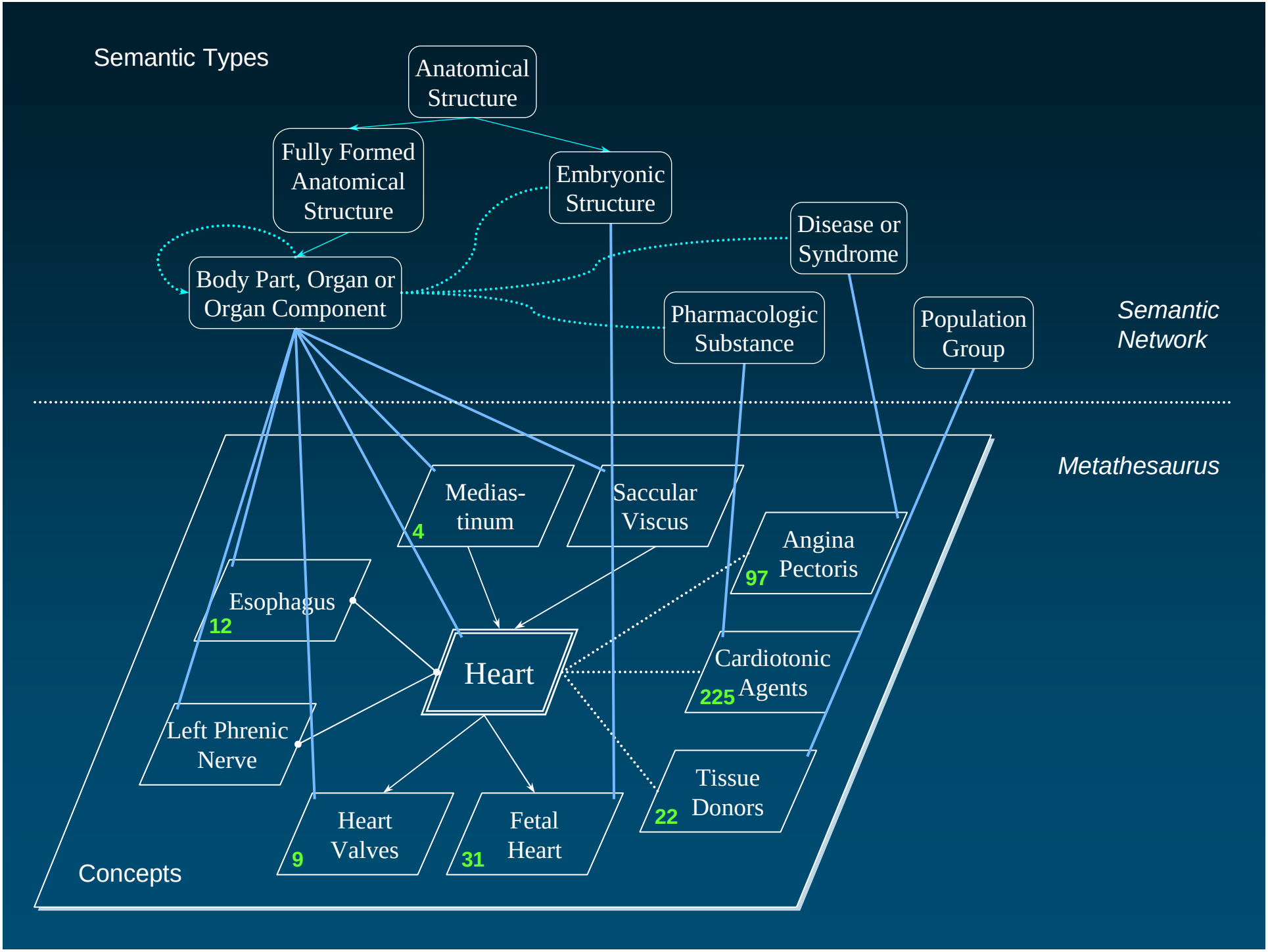
Symbolic relationships Type

- Hierarchical
  - Parent / Child — **PAR/CHD**
  - Broader / Narrower than — **RB/RN**
- Derived from hierarchies
  - Siblings (children of parents) — **SIB**
- Associative
  - Other — **RO**
- Various flavors of near-synonymy
  - Similar — **RL**
  - Source asserted synonymy — **SY**
  - Possible synonymy — **RQ**



Symbolic relationships Attribute

- Hierarchical
  - isa (is-a-kind-of)
  - part-of
- Associative
  - location-of
  - caused-by
  - treats
  - …
- Cross-references (mapping)


Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Disease or Syndrome
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
Semantic Network
Metathesaurus
Medias-tinum
4
Saccular Viscus
Angina Pectoris
97
Esophagus
12
Heart
Cardiotonic Agents
225
Left Phrenic Nerve
Tissue Donors
22
Heart Valves
9
Fetal Heart
31
Concepts

UMLS Semantic Network

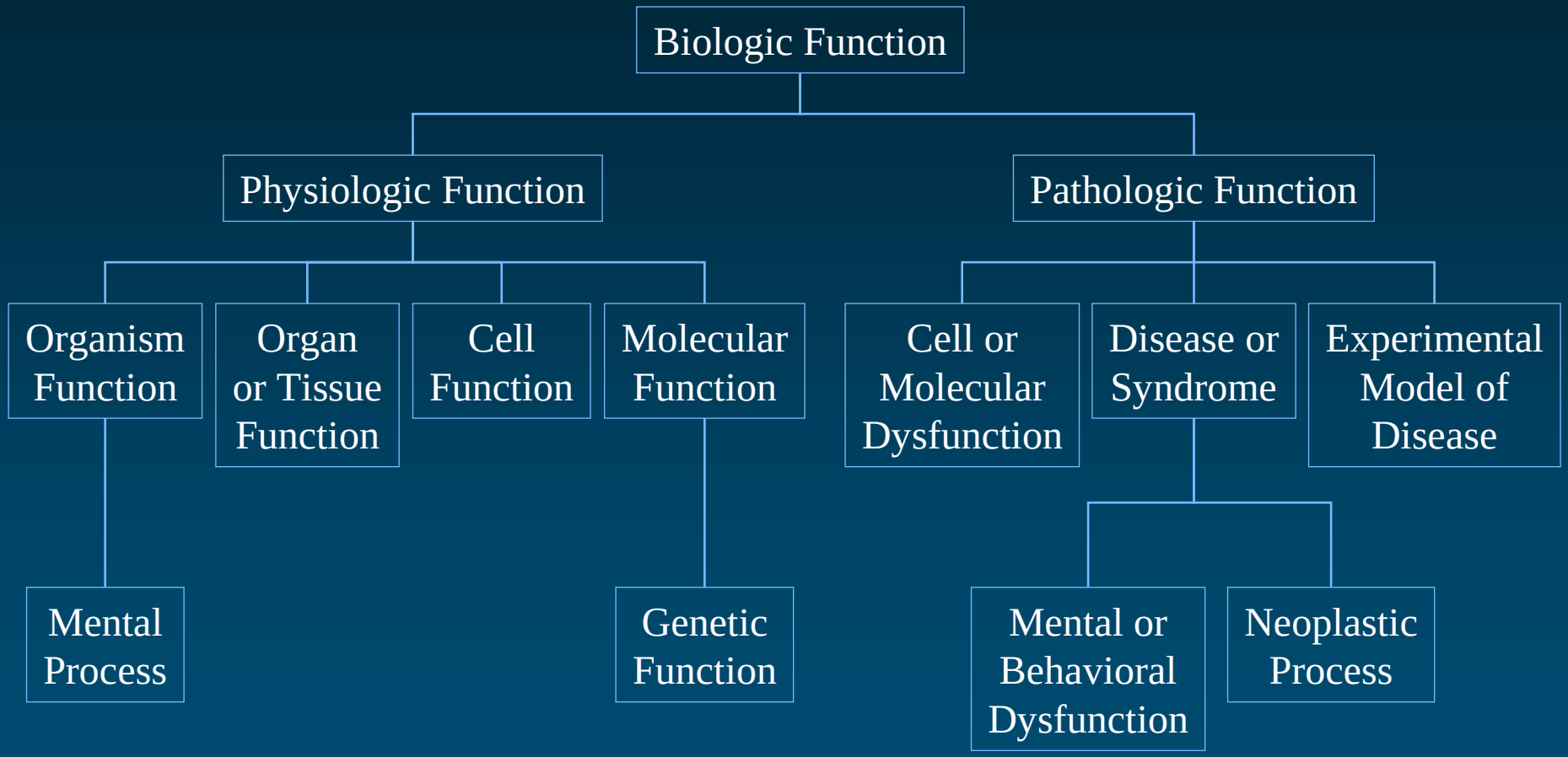
Semantic Network

- Semantic types (135)
  - tree structure
  - 2 major hierarchies
    - Entity
      - Physical Object
      - Conceptual Entity
    - Event
      - Activity
      - Phenomenon or Process

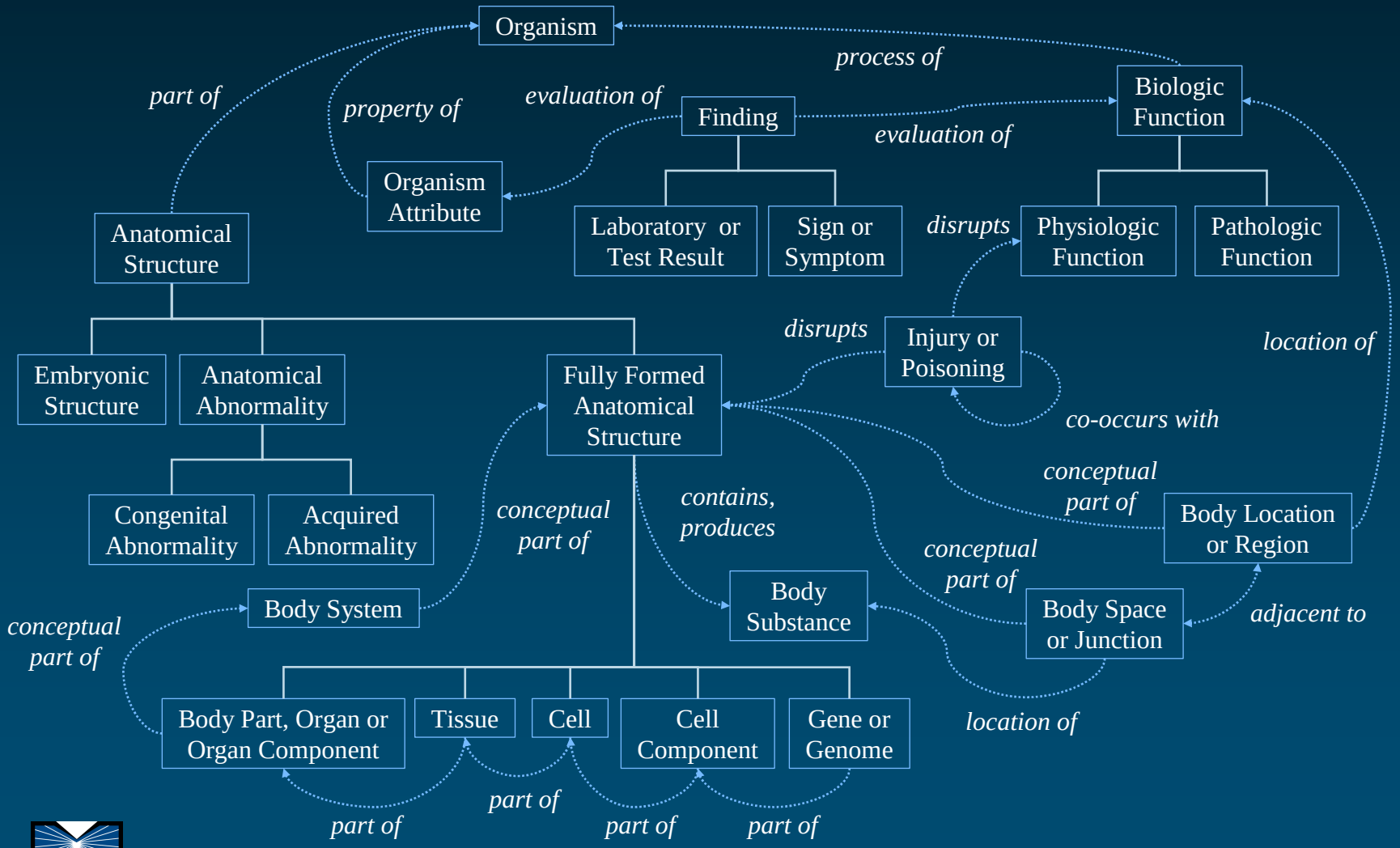
Semantic Network

- Semantic network relationships (54)
  - hierarchical (isa = is a kind of)
    - among types
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - among relations
      - treats *isa* affects
  - non-hierarchical
    - Sign or Symptom *diagnoses* Pathologic Function
    - Pharmacologic Substance *treats* Pathologic Function

“Biologic Function” hierarchy (isa)



Associative (non-isa) relationships

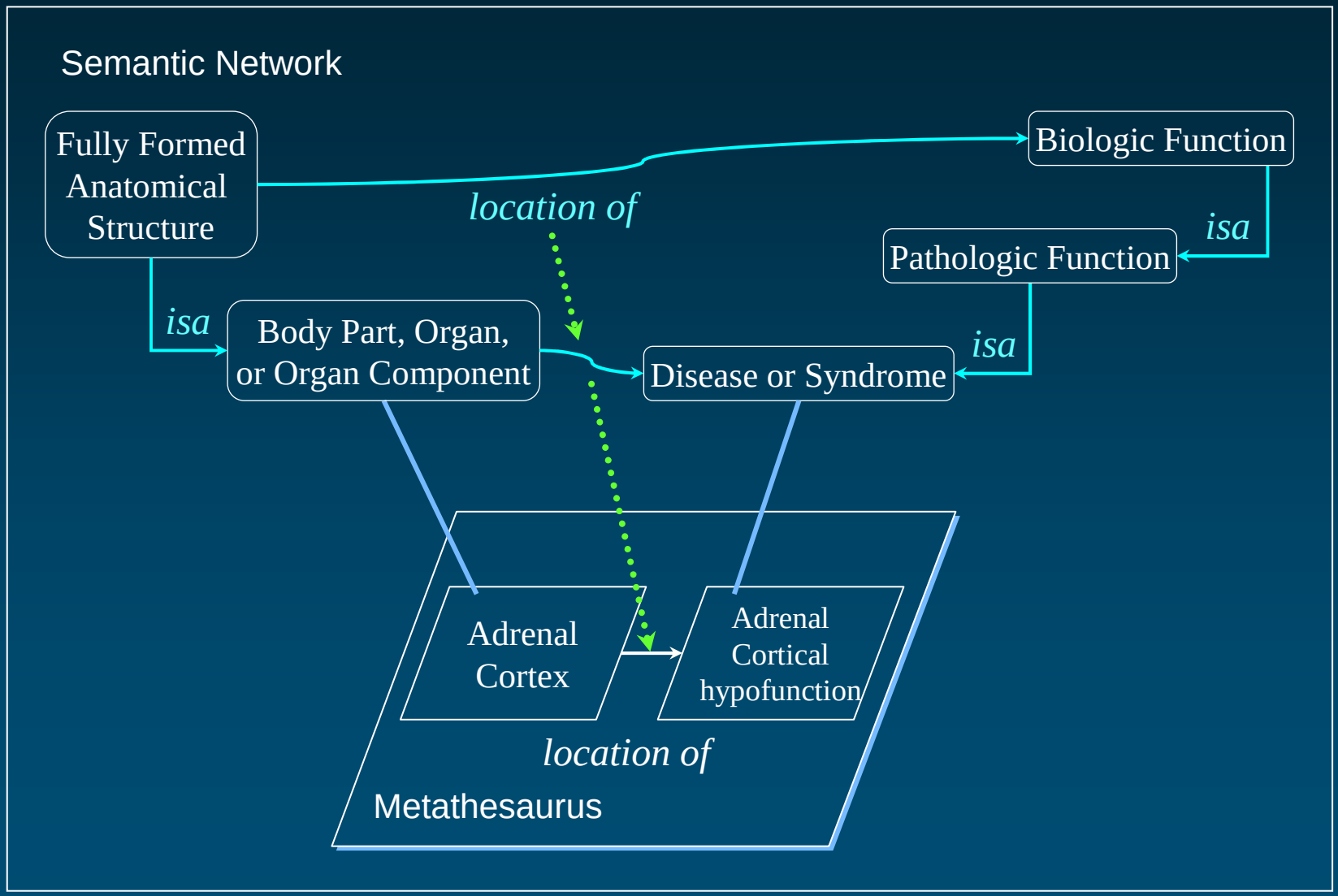


Why a semantic network?

- Semantic Types serve as high level categories assigned to Metathesaurus concepts, *independently of their position in a hierarchy*

- A relationship between 2 Semantic Types (ST) is a possible link between 2 concepts that have been assigned to those STs
  - The relationship may or may not hold at the concept level
  - Other relationships may apply at the concept level

Relationships can inherit semantics



SPECIALIST Lexicon and lexical tools

SPECIALIST Lexicon

- Content
  - English lexicon
  - Many words from the biomedical domain
- 200,000+ lexical items
- Word properties
  - morphology
  - orthography
  - syntax
- Used by the lexical tools

Morphology

- Inflection
  - noun — nucleus, nuclei
  - verb — cauterize, cauterizes, cauterized, cauterizing
  - adjective — red, redder, reddest
- Derivation
  - verb ⟺ noun — cauterize -- cauterization
  - adjective ⟺ noun — red -- redness

Orthography

- Spelling variants
  - oe/e — oesophagus - esophagus
  - ae/e — anaemia - anemia
  - ise/ize — cauterise - cauterize
  - genitive mark — Addison's disease
Addison disease
Addisons disease

Syntax

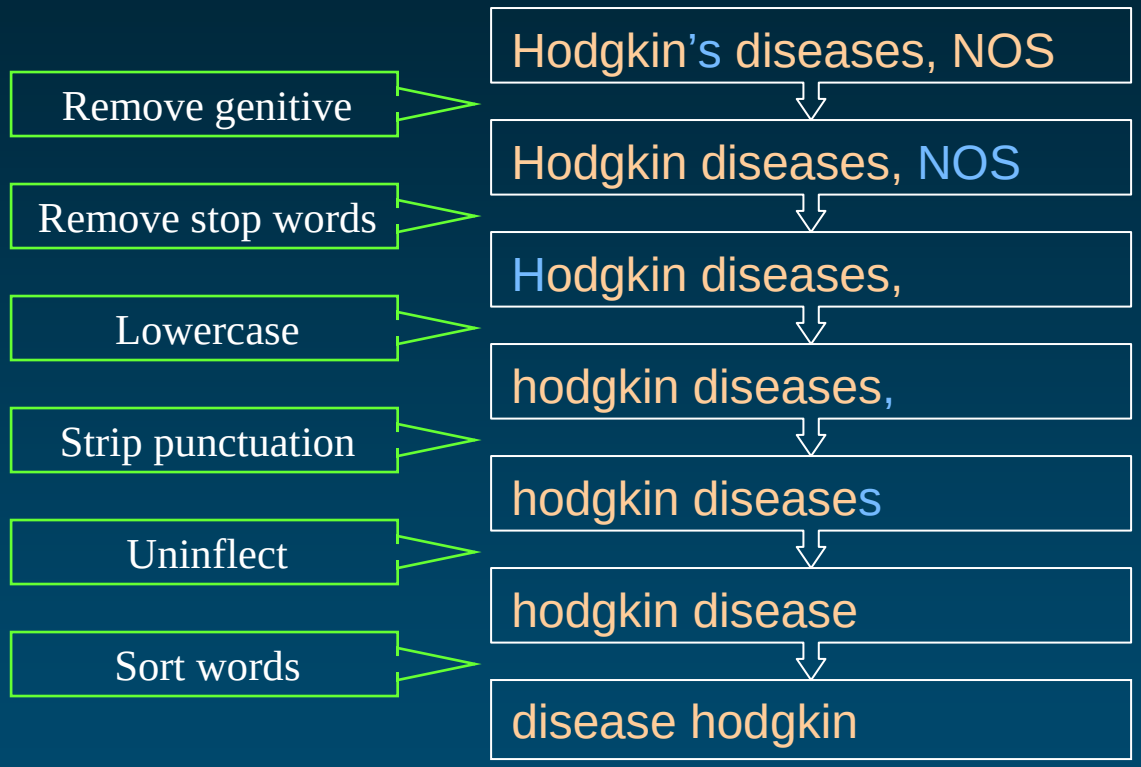
- Complementation
  - verbs
    - intransitive — I'll treat.
    - transitive — He treated the patient.
    - ditransitive — He treated the patient with a drug.
  - nouns
    - prepositional phrase

      Valve of coronary sinus
- Position for adjectives

Lexical tools

- To manage lexical variation in biomedical terminologies
- Major tools
  - Normalization
  - Indexes
  - Lexical Variant Generation program (lvg)
- Based on the SPECIALIST Lexicon
- Used by noun phrase extractors, search engines

Normalization



Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

Normalization Applications

- Model for lexical resemblance
- Help find lexical variants for a term
  - Terms that normalize the same usually share the same LUI
- Help find candidates to synonymy among terms
- Help map input terms to UMLS concepts

Indexes

- Word index
  - word to Metathesaurus strings
  - one word index per language
- Normalized word index
  - normalized word to Metathesaurus strings
  - English only
- Normalized string index
  - normalized term to Metathesaurus strings
  - English only

Lexical Variant Generation program

- Tool for specialists (linguists)
- Performs atomic lexical transformations
  - generating inflectional variants
  - lowercase
  - …
- Performs sequences of atomic transformations
  - a specialized sequence of transformations provides the normalized form of a term (the *norm* program)

UMLS in action
MetaMap

MetaMap Motivation

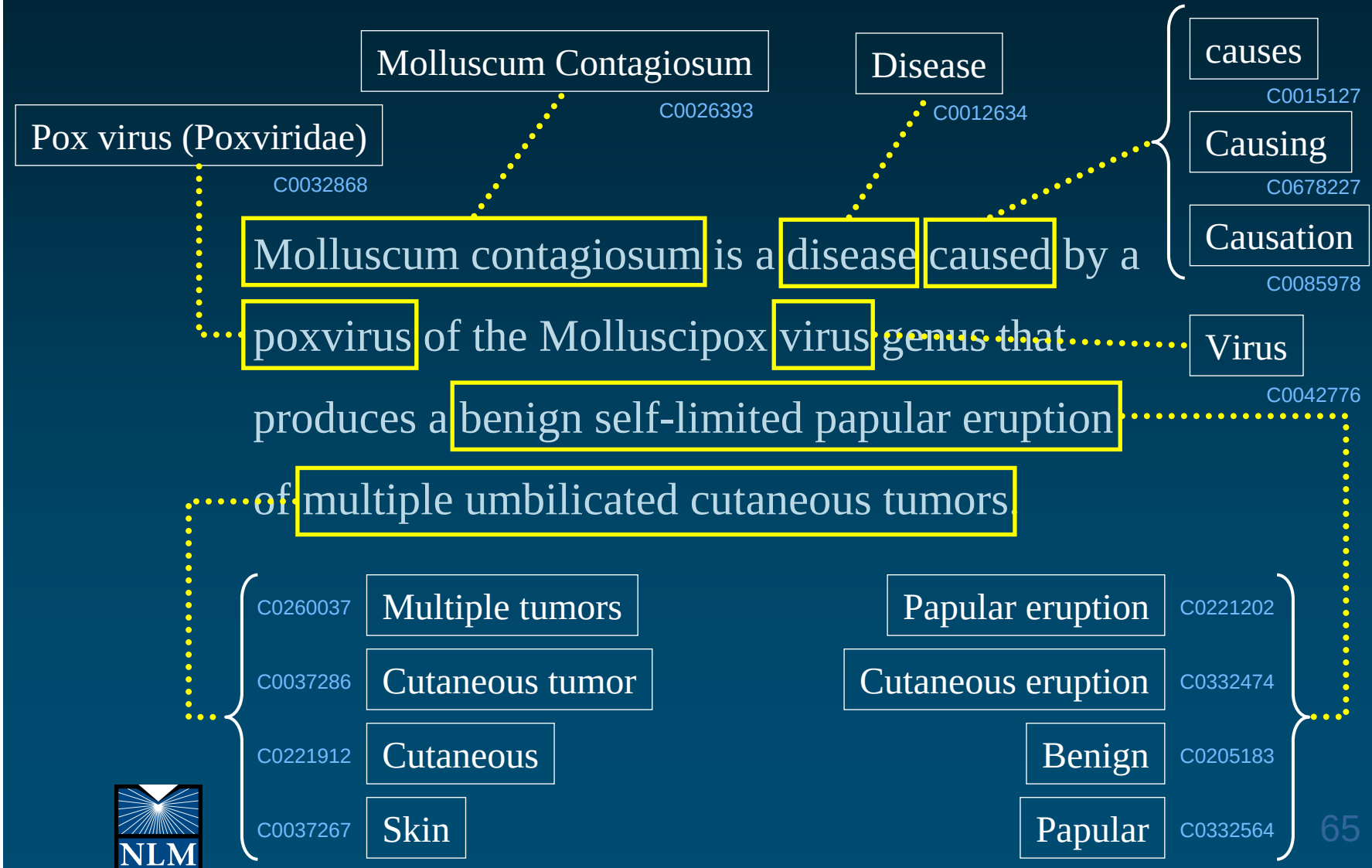
[Aronson, *AMIA*, 2001]

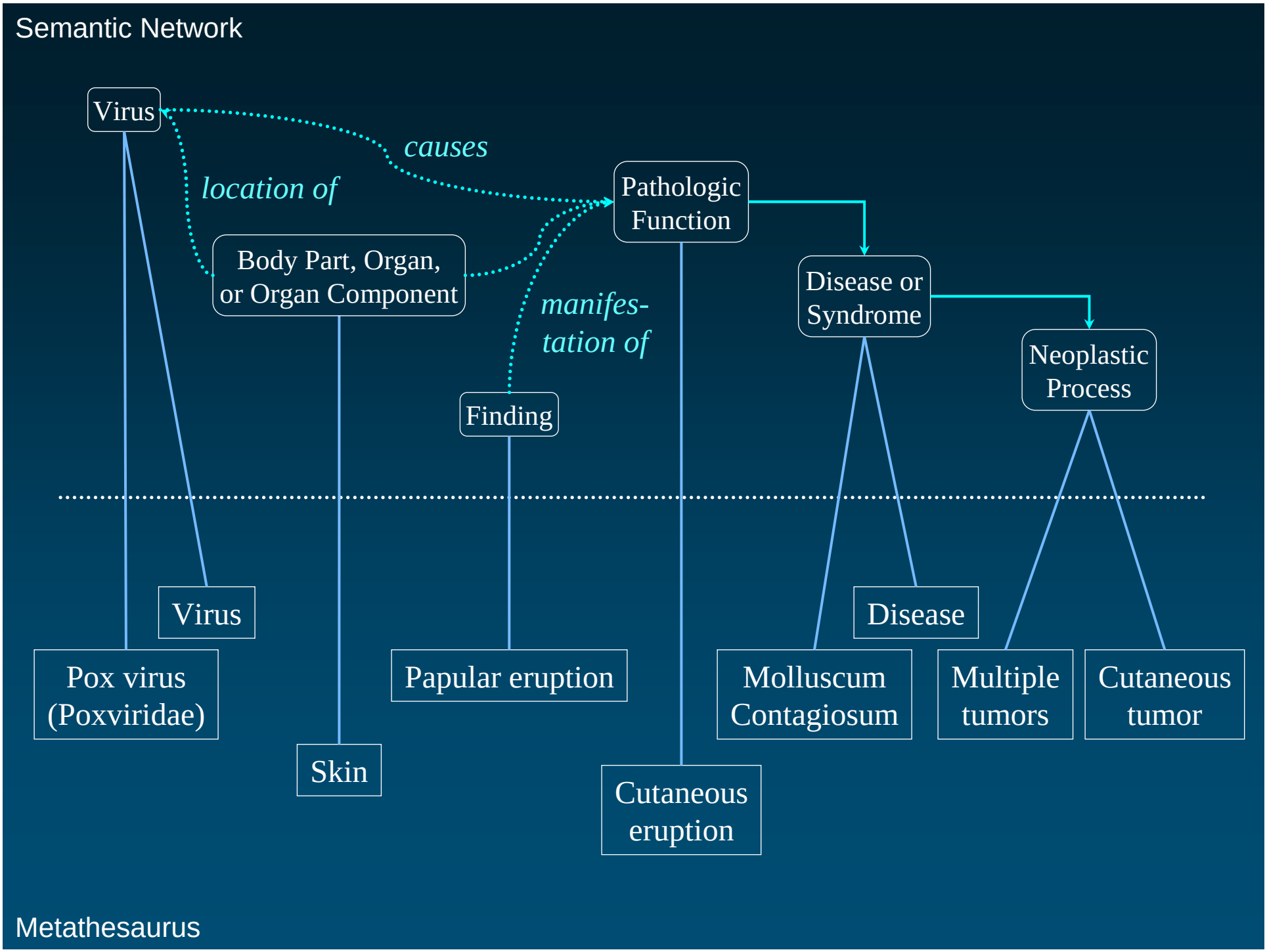
- Term extraction
  - Identifying UMLS concepts from text
- Usage
  - Information indexing and retrieval
  - Knowledge extraction / discovery
  - Semantic interpretation
- Characteristics
  - Linguistic approach
  - Based on UMLS knowledge sources

MetaMap Methods

- Parsing
  - Shallow syntactic analysis
  - SPECIALIST lexicon
  - Xerox part-of-speech tagger
- Variant generation
- Candidate retrieval
  - Retrieve candidate terms containing at least one variant
- Candidate evaluation
  - Rank candidate terms with respect to closeness to input text (centrality, variation, coverage, and cohesiveness)

MetaMap Example

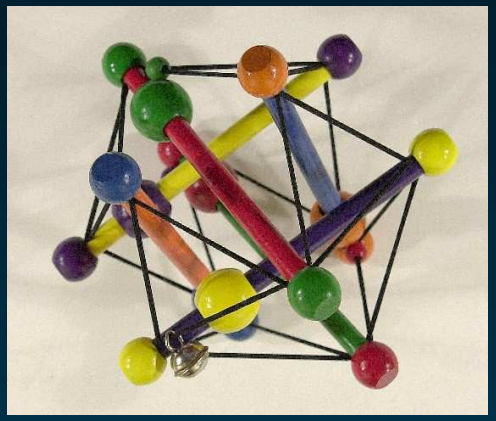



Semantic Network
Virus
causes
location of
Pathologic Function
Body Part, Organ, or Organ Component
manifes-tation of
Disease or Syndrome
Neoplastic Process
Finding
Virus
Disease
Pox virus (Poxviridae)
Papular eruption
Molluscum Contagiosum
Multiple tumors
Cutaneous tumor
Skin
Cutaneous eruption
Metathesaurus

Using MetaMap MMTx

- Requires UMLS license
- Local implementation (Java-based)
- Provides
  - Stand-alone application
  - API for integrating in other applications

http://mmtx.nlm.nih.gov



Medical Ontology Research

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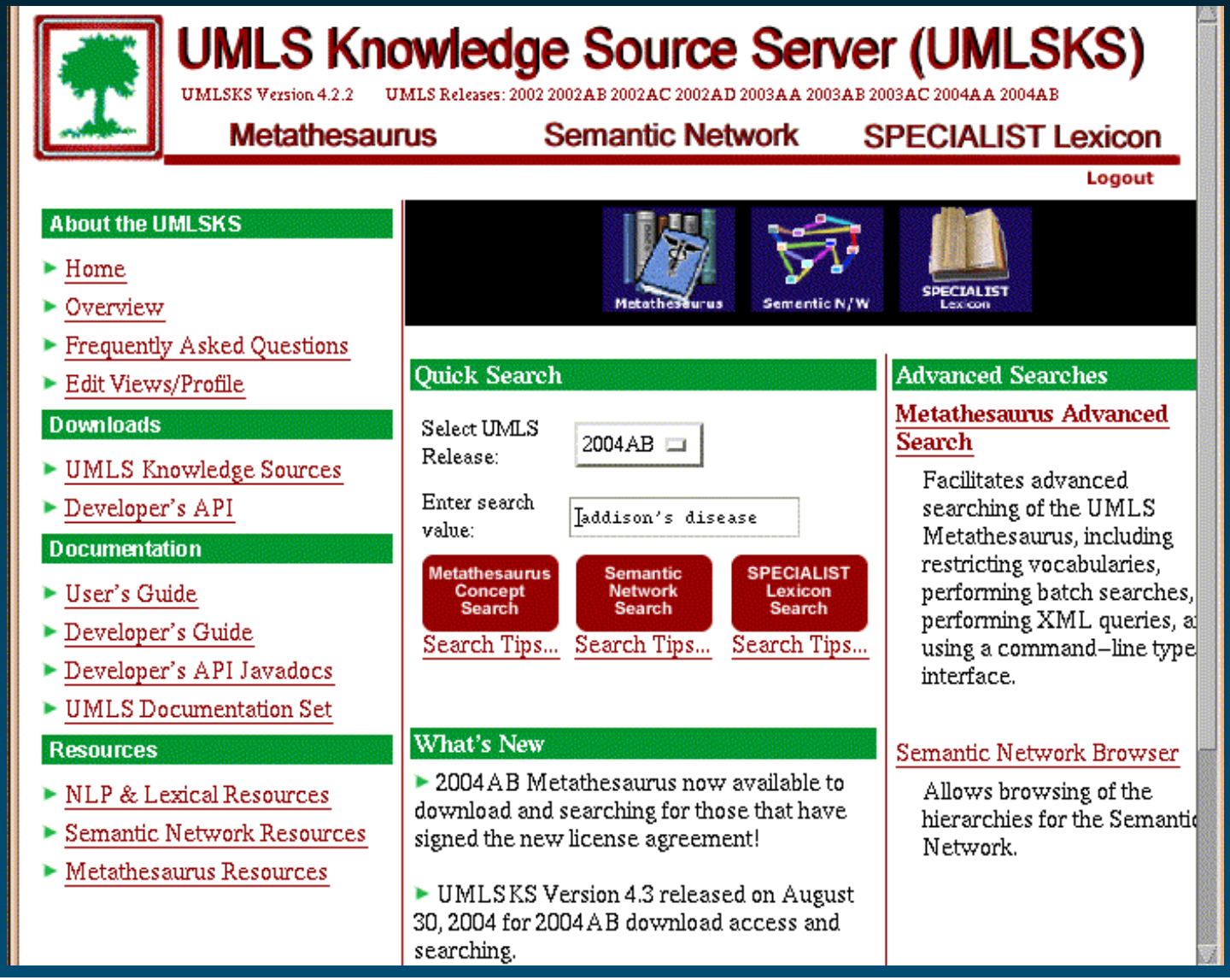
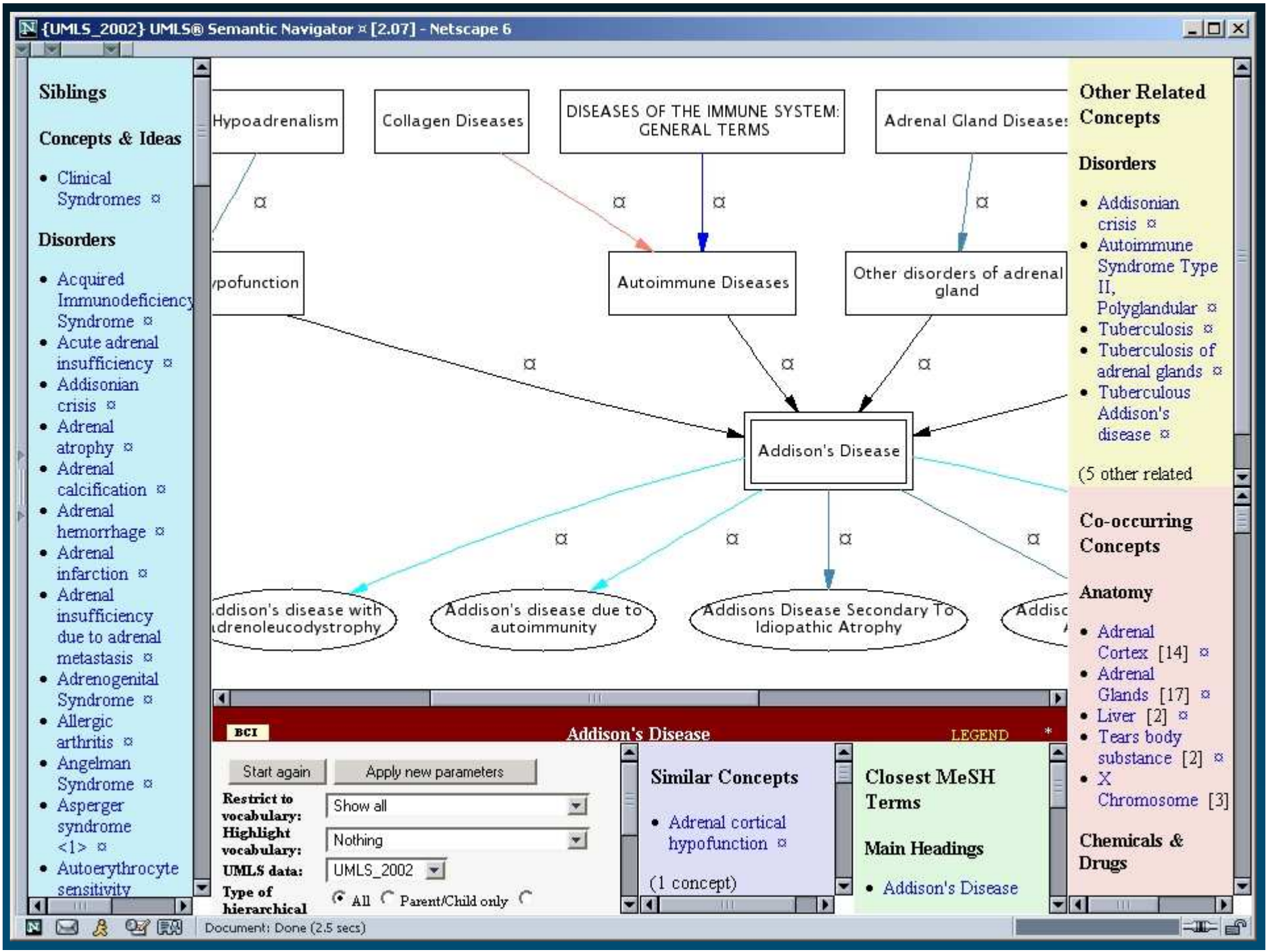
Appendix

Knowledge Source Server
Web Interface

http://umlsks.nlm.nih.gov

UMLS Knowledge Source Server Home Page




{UMLS_2002} UMLS® Semantic Navigator ¤ [2.07] - Netscape 6
Siblings
Concepts & Ideas
• Clinical Syndromes ¤
Disorders
• Acquired Immunodeficiency Syndrome ¤
• Acute adrenal insufficiency ¤
• Addisonian crisis ¤
• Adrenal atrophy ¤
• Adrenal calcification ¤
• Adrenal hemorrhage ¤
• Adrenal infarction ¤
• Adrenal insufficiency due to adrenal metastasis ¤
• Adrenogenital Syndrome ¤
• Allergic arthritis ¤
• Angelman Syndrome ¤
• Asperger syndrome <1> ¤
• Autoerythrocyte sensitivity
Hypoadrenalism
Collagen Diseases
DISEASES OF THE IMMUNE SYSTEM: GENERAL TERMS
Adrenal Gland Diseases
ypofunction
Autoimmune Diseases
Other disorders of adrenal gland
Addison's Disease
ddison's disease with drenoleucodystrophy
Addison's disease due to autoimmunity
Addisons Disease Secondary To Idiopathic Atrophy
Other Related Concepts
Disorders
• Addisonian crisis ¤
• Autoimmune Syndrome Type II, Polyglandular ¤
• Tuberculosis ¤
• Tuberculosis of adrenal glands ¤
• Tuberculous Addison's disease ¤
(5 other related
Co-occurring Concepts
Anatomy
• Adrenal Cortex [14] ¤
• Adrenal Glands [17] ¤
• Liver [2] ¤
• Tears body substance [2] ¤
• X Chromosome [3]
Chemicals & Drugs
BCI
Addison's Disease
LEGEND
Start again
Apply new parameters
Restrict to vocabulary:
Show all
Highlight vocabulary:
Nothing
UMLS data:
UMLS_2002
Type of hierarchical
All
Parent/Child only
Similar Concepts
• Adrenal cortical hypofunction ¤
(1 concept)
Closest MeSH Terms
Main Headings
• Addison's Disease
Document: Done (2.5 secs)

Knowledge Source Server
Application Programming Interface

UMLSKS API basics

- Remote server at NLM
- Local application connected through

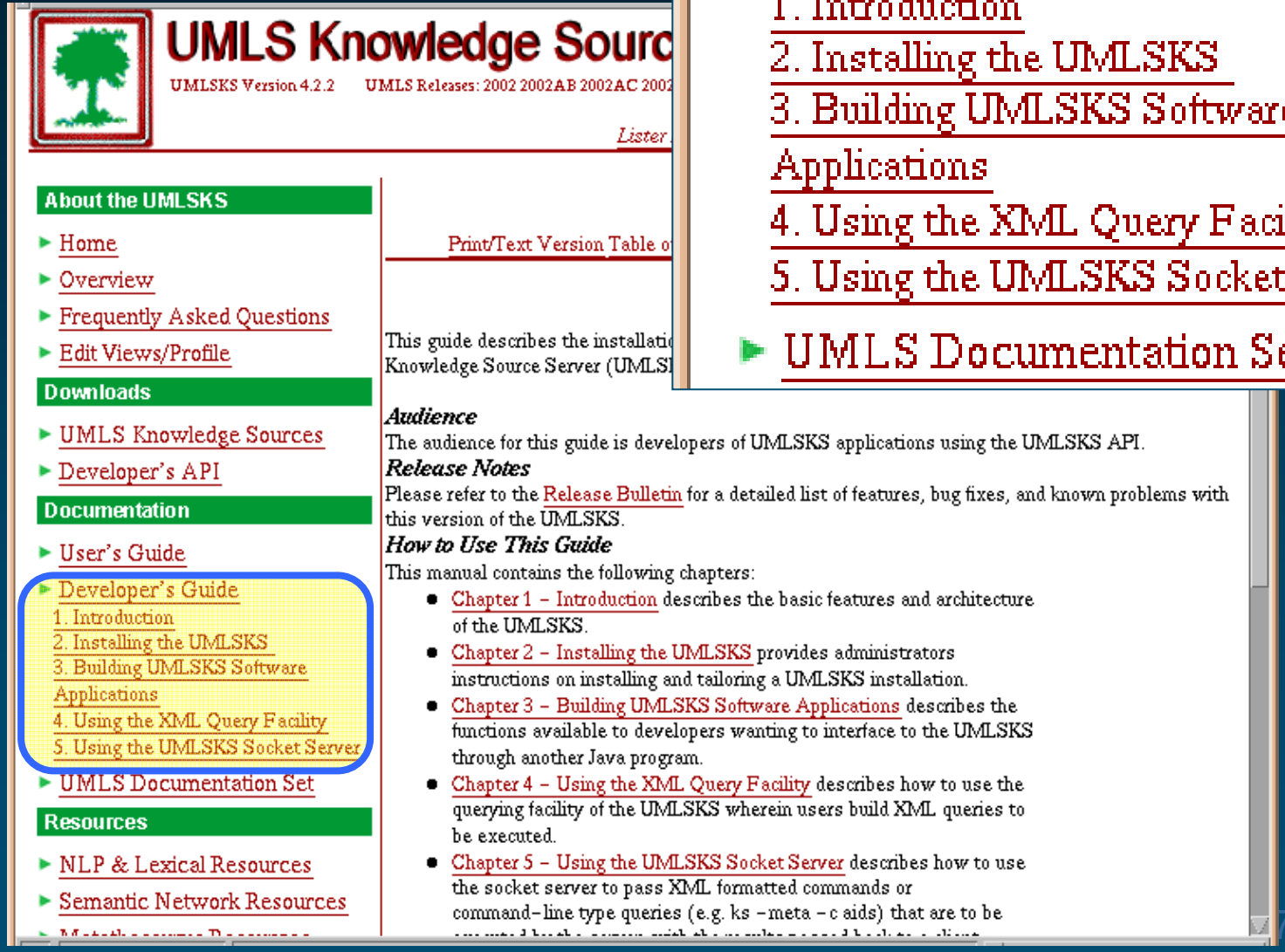
Java RMI

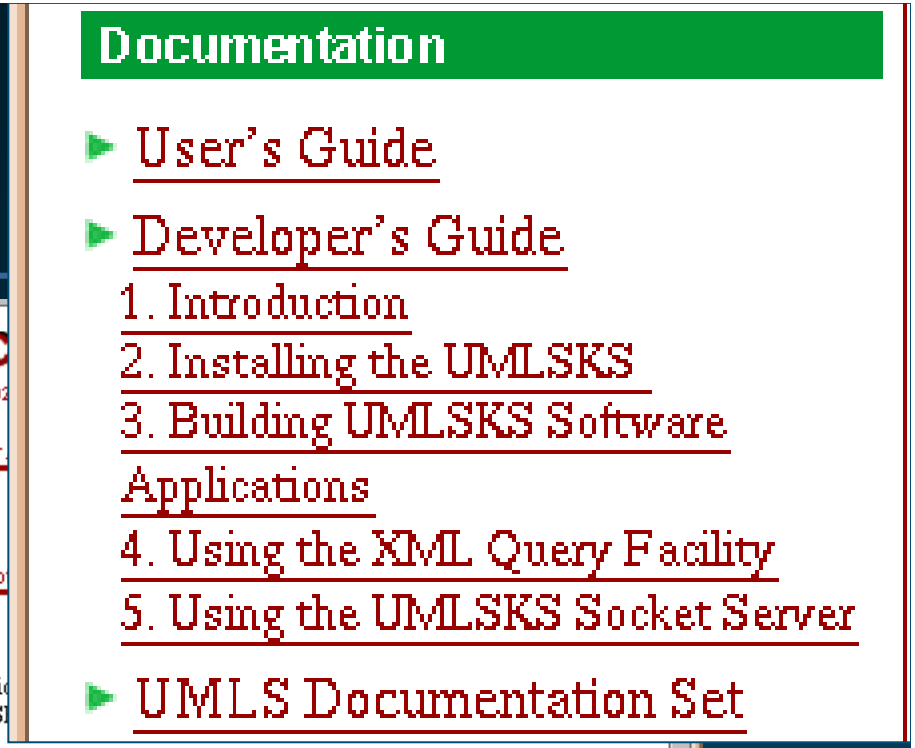
- Java-based applications
- Developer's Guide: Chapter 3
- Set of Java classes (part of the UMLSKS API download)
- Detailed *Javadoc* documentation online and with API download

TCP/IP socket

- XML-based queries
- Developer's Guide: Chapter 5
- XML schema
- Socket server
  - Host: umlsks.nlm.nih.gov
  - Port: 8042

Developer's Guide





MetamorphoSys

What is MetamorphoSys?

- Tool distributed with the UMLS
- Multi-platform Java software
- The UMLS installation and customization wizard
  - Installs Knowledge Sources to local storage
  - Subsets and customizes a local Metathesaurus

Why use MetamorphoSys?

Customize the Metathesaurus

- To remove terminology that is unhelpful, or even harmful, to your needs and purposes
- To comply with terms of license agreement

Changing Default Settings

- To alter the preferred name
- To alter suppressibility of specific source term types

Bibliography

UMLS documentation and support

- UMLS homepage http://umlsinfo.nlm.nih.gov/
  - with links to all other UMLS information
- UMLSKS homepage http://umlsks.nlm.nih.gov/
  - with links to the User's and Developer's guides
- Email address for support custserv@nlm.nih.gov

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  - Humphreys, B. L., Lindberg, D. A., Schoolman, H. M., & Barnett, G. O. (1998). The Unified Medical Language System: an informatics research collaboration. *J Am Med Inform Assoc*, *5*(1), 1-11.
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  - McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, *34*(1-2), 193-201.
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- Comprehensive bibliography 1986-96

  http://www.nlm.nih.gov/pubs/cbm/umlscbm.html